Progressive hemifacial atrophy with agenesis of the head of the caudate nucleus

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Abstract

We describe a woman with right hemifacial atrophy, a high palate, partial left motor seizures, and mild atrophy of the left arm. CT scan showed asymmetrical lateral ventricles and MRI (magnetic resonance imaging) showed atrophy of the right cerebral hemisphere and agenesis of the head of the right caudate nucleus. To our knowledge, this is the first report of Parry-Romberg syndrome associated with structural abnormalities of the basal nuclei documented by MRI. We suggest that a neurovascular aetiology can explain the spectrum of segmental defects associated with hemifacial atrophy.

(J Med Genet 1994;31:969-971)

Parry-Romberg syndrome or hemifacial atrophy is an uncommon and poorly understood condition manifested by progressive hemifacial atrophy of skin, soft tissue, and bone within one or more trigeminal nerve dermatomes. The symptoms usually begin in the first or second decades[1] sometimes associated with contralateral partial motor seizures,[2 3] trigeminal neuralgia,[4] mastigatory spasms,[5-9] hemiplegic migraine,[10] and cerebral or cerebellar malformations.[2 3 11 12] Other abnormalities associated with hemifacial atrophy are atrophy of the trunk and extremities,[13-16] scleroderma,[6 14 17-19] systemic lupus erythematosus,[18] ocular defects,[4 20] and Poland syndrome.[21]

We describe a patient with Parry-Romberg disease associated with agenesis of the head of the right caudate nucleus.

Case report

A 31 year old woman was referred to the Department of Neurology, when she developed partial motor seizures of the left arm. She had right facial atrophy and hyperpigmentation over the second and third divisions of the trigeminal nerve (fig 1), a high palate, mild atrophy of the right side of the tongue, and mild atrophy of the left arm. Her pectoral muscles and her fingers were normal. Since her teens she had been aware of progressive atrophy of the right side of her face. She reported two episodes of spasms of the muscles of the right and left jaw, lasting for a few seconds and precipitated by the movement of the jaw while eating, when she was 29 years old.

Her neurological examination showed brisk reflexes in the left arm. Ocular movements, pupillary reactions, and the ocular fundus were normal. Sensation was normal in all divisions of both fifth cranial nerves, trunk, arms, and legs.

The electroencephalogram showed generalised low voltage with delta waves in the right temporal region. The CT scan showed asymmetrical lateral ventricles and enlargement of the right-central sulcus, suggesting partial atrophy of the right hemisphere. MRI showed atrophy of the right cerebral hemisphere, agenesis of the head of the right caudate nucleus, and partial absence of the right putamen (fig 2A and B). Antinuclear antibodies were absent.

The family history was unremarkable. The mother and the father were 24 and 29 years old respectively at the time of the patient's birth and were not consanguineous. The patient's birth and previous medical history were uneventful.

Discussion

This patient has several abnormalities usually described in Parry-Romberg syndrome: hemifacial atrophy, contralateral partial motor seizures, contralateral atrophy of the arm, and mastigatory spasms. The agenesis of the head of the right caudate nucleus has not previously

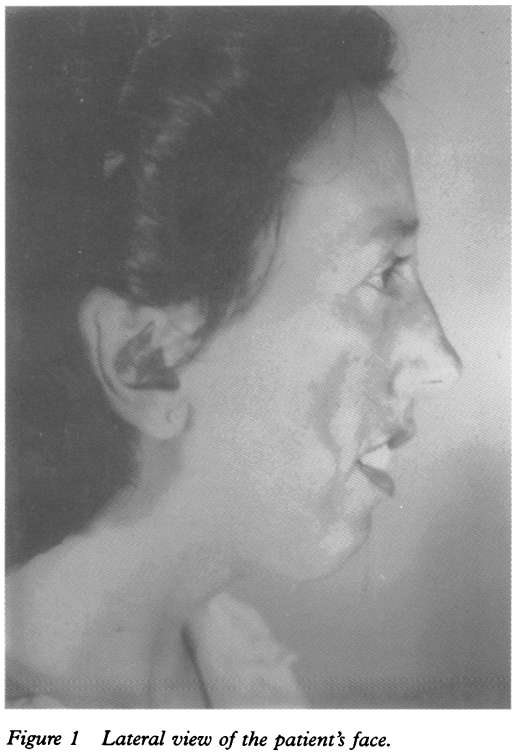

Figure 1 Lateral view of the patient's face.

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Received 16 November 1993.
Revised version accepted for publication 22 July 1994.

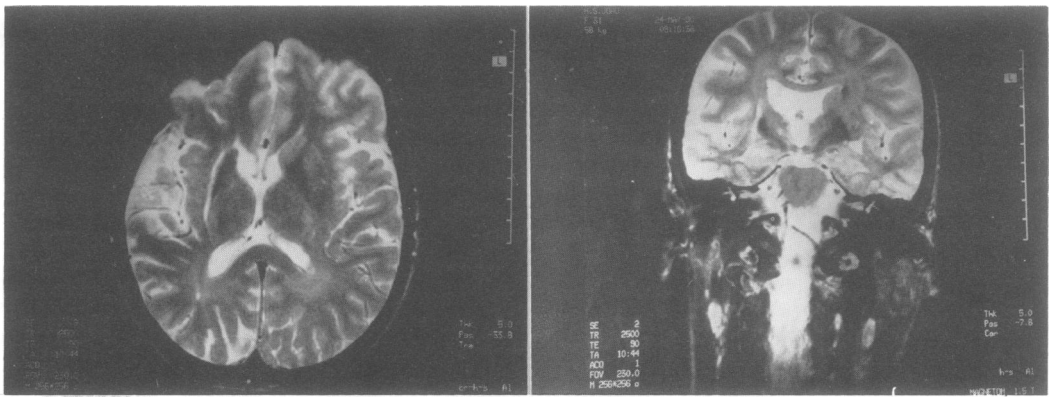

Figure 2 Axial (left) and coronal (right) T2 weighted images (2500/90) showing atrophy of the right cerebral hemisphere, agenesis of the head of the right caudate nucleus, and partial agenesis of the right putamen.

been reported and enlarges the spectrum of structural defects described in this disorder.

There are several published reports that support the hypothesis of a neurovascular aetiology in hemifacial atrophy. Dintiman *et al*[21] described a patient with Parry-Romberg disease associated with a contralateral Poland anomaly. This defect can be caused by a subclavian artery supply disruption.[22,23] A case of atrophy of the rhomboid muscles associated with hemifacial atrophy was reported by Zafarulla[16] and there are other reports showing the association of an abnormal arterial supply with isolated or familial cases of muscular agenesis.[24] Hirata *et al*[13] reported a case of crossed total hemiatrophy involving the right side of the face and the left side of the trunk and extremities in a patient with a right precentral to central arteriovenous malformation. Hemiplegic migraine has also been reported in patients with Parry-Romberg disease.[10] These reports document the association of Parry-Romberg disease with several defects of vascular supply.

Partial motor seizures, occurring contralaterally to the side of the facial atrophy, have also been described in Parry-Romberg patients. These seizures are probably caused by brain malformations[2,3]; the results of the EEG and neuroradiological investigations performed in our patient support this hypothesis.

Mastigatory spasms occur frequently in patients with Parry-Romberg disease.[5–9] A structural defect of the basal nuclei can explain these involuntary movements. The occurrence of mastigatory spasms in our patient and the results of MRI imaging, showing complete agenesis of the head of the right caudate nucleus and partial absence of the putamen, lead us to speculate about the relationship between a structural defect of the basal nuclei and the occurrence of mastigatory spasms in facial hemiatrophy. Although routine neuroradiological examination has not been systematically performed in Parry-Romberg disease, several authors have reported abnormal CT scans in patients with this disorder.[2,11,12] However, since structural abnormalities of the basal ganglia are easily missed on CT scanning, this might explain why other authors did not find this abnormality in patients with Parry-Romberg disease suffering from mastigatory spasms.

Garcher *et al*[25] suggested that sympathetic overactivity owing to a dysfunction in the mesencephalic area or in the superior cervical sympathetic ganglion might account for the association of Fuch and Horner's syndromes with hemifacial atrophy. A disruption of vascular supply caused by abnormal vessels or by sympathetic overactivity is a plausible explanation for the association of Parry-Romberg disease with hemiplegic migraine,[10] with several segmental defects such as atrophy of the trunk and extremities,[13–16] and with Poland syndrome.[21] Experimental evidence of sympathetic dysfunction in progressive facial hemiatrophy was recently provided by Resende *et al.*[26]

The coexistence of Parry-Romberg disease with scleroderma,[14,17,18] systemic lupus erythematosus,[18] and uveitis[20] is well known. However, the relationship between these autoimmune diseases and hemifacial atrophy is not clear. The results of microscopic studies in patients with Parry-Romberg disease show abnormal lymphocytic infiltration of the vascular endothelium and basement membranes, suggesting chronic cell mediated vascular injury.[27] Biopsy specimens show the same kind of plasma and lymphocytic cells as found in patients with scleroderma.[28,29] These findings suggest that an immune reaction (either cellular or humoral) can explain the clinical evolution of hemifacial atrophy.

It is difficult to explain the coexistence of disruptive defects, autoimmune diseases, and sympathetic overactivity in patients with hemifacial atrophy. However, long standing adrenergic or noradrenergic activity could lead to an exaggerated constriction of blood vessels causing necrosis or an abnormal development of several body segments, and, secondarily, to a cascade of immunological reactions.

Viewing the neurological abnormalities frequently found in patients with facial hemiatrophy (seizures and mastigatory spasms) we recommend routine CT scan and MRI examination of those patients, to detect any anatomical defects of the cerebral hemispheres and basal ganglia.

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